

5

10

25

- 1. A method for detecting the presence of or predisposition to an ectodermal disorder comprising the steps of:
  - (a) detecting the presence of a TAJ gene or gene product in a cell; and
- (b) correlating the presence of the TAJ gene or gene product with a presence of or predisposition to an ectodermal disorder.
- The 2. A method according to claim 1, wherein the detecting step comprises detecting a TAJ gene.
- The 3. A method according to claim 1, wherein the detecting step comprises detecting a TAJ gene transcript.
- The 4. A method according to claim 1, wherein the detecting step comprises detecting a TAJ protein.
- 5. A-method according to claim 1, wherein the detecting step is performed inferentially by determining a diagnostic sequence of the TAJ gene or gene product in the individual.
- 6. A method according to claim 1, wherein the TAJ gene or gene product is a variant correlated with the presence of or predisposition to an ectodermal disorder.
- 7. Amethod according to claim 1, wherein the ectodermal disorder is an ectodermal dysplasia syndrome.
- 8. A method according to claim 1, wherein the ectodermal disorder is an ectodermal dysplasia syndrome and the syndrome is Clouston syndrome.
- 9. A method for modulating the functional expression of a TAJ gene or gene product in a cell comprising the step(s) of:

  contacting a cell with an agent which specifically binds and modulates the functional

19

5

- (a) the cell is an ectodermal cell; or
- (b) the cell is a germ cell which gives rise to progeny ectodermal cells and detecting the functional expression of the TAJ gene or gene product in the progeny cells.
- 10. A method according to claim 9, wherein the cell is in situ.
- 11. A method according to claim 9, wherein the cell is ex situ.
- 10 12. A method according to claim 9, wherein the contacting step reduces the functional expression of the TAJ gene or gene product.
  - 13. A method according to claim 9, wherein the agent is an antibody which specifically binds a TAJ protein.
  - 14. A method according to claim 9, wherein the agent is an intrabody which specifically binds a TAJ protein.
  - The 15. A method according to claim 9, wherein the agent is an agonist or antagonist of a TAJ protein.
  - 16. A method according to claim  $\theta$ , wherein the agent is an antisense oligonucleotide which specifically binds a TAJ gene transcript.
- 25 17. A method according to claim 9, wherein the agent is an oligonucleotide which specifically binds a TAJ gene.
  - 18. A method according to claim 9, wherein the agent is an oligonucleotide which specifically binds a TAJ gene, whereby the gene is changed to a different TAJ gene.
  - 19. A method according to claim 9, wherein the agent is an oligonucleotide which

20

The state of the s

30

5

10

specifically binds a TAJ gene, whereby the gene is changed from a TAJ gene correlated with a presence of or predisposition to an ectodermal disorder to a different TAJ gene not so correlated.

- 20. A method according to claim 9, wherein the agent is an oligonucleotide which specifically binds a TAJ gene, whereby the gene is changed from a TAJ gene correlated with a presence of or predisposition to an ectodermal disorder to a different TAJ gene not so correlated, wherein the ectodermal disorder is an ectodermal dysplasia syndrome.
- 21. A method according to claim 9, wherein the agent is an oligonucleotide which specifically binds a TAJ gene, whereby the gene is changed from a TAJ gene correlated with a presence of or predisposition to an ectodermal disorder to a different TAJ gene not so correlated, wherein the ectodermal disorder is an ectodermal dysplasia syndrome and the syndrome is Clouston syndrome.